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From Respiratory Distress to Diagnosis of Amyotrophic Lateral Sclerosis

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Abstract

Amyotrophic lateral sclerosis (ALS) is a progressive neurodegenerative disease of motor neurons, which results in weakness and atrophy of voluntary skeletal muscles. ALS is the most common form of motor neuron disease (MND). It is currently incurable and treatment is largely limited to supportive care. Family history is associated with an increased risk of ALS. The disease usually does not affect cognitive abilities, but causes muscle weakness. Respiratory function in the early stages of the disease is generally normal, shortness of breath occurs as the disease progresses. This paper has been submitted about ALS patient who was diagnosed and treated in intensive care unit followed by severe respiratory failure.

Keywords: Amyotrophic lateral sclerosis, respiratory failure

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Introduction

ALS is an illness which the other names are Charcot's disease or Lou Gehrig's disease. ALS is a motor neuron disease caused by progressive degeneration of motor neurons in medulla spinalis and brain stem. At the present time, there are about 25,000 patients with ALS in the USA, with an average starting age of 55 years. The incidence and prevalence of ALS are 1-2 and 4-6 per 100,000 every year, respectively, with a lifetime ALS risk of 1/600 to 1/1000ALS's presentation, a variety of courses and progress. Because the causes and mechanism of the disease is not known clearly there is not any effective treatment [1]. Disease presents with muscle weakness and atrophy, mental and memory functions remain intact. The disease is fatal within nearly three years after diagnosis has been made. Death usually occurs due to respiratory failure [2]. The patients are usually diagnosed with muscle weakness. In some cases it can be diagnosed after development of respiratory distress. In this study we present a patient who was followed in intensive care due to respiratory distress and was diagnosed with ALS in the light of the known literature.

Case report

Fifty nine years old female patient admitted to our Intensive Care Unit (ICU) because of respiratory distress after posterior vertebral instrumentation surgery. Patient had lower extremity weakness and hoarseness for nearly a year. Patient was operated one week earlier because of lumbar disc herniation and spinal stenosis. She had respiratory distress after operation and her status worsened thus she was intubated and admitted to ICU. The patient had a history of hypertension.

Thorax CT scan and MRI showed minimal bilateral pleural effusion which did not explain her severe clinical condition. Possible cardiac etiology was ruled out with normal electrocardiography and echocardiography. Cerebral CT and MRI scans were also normal. While the patient was mentally cooperated and had purposeful motor function after sedation was ceased; she experienced respiratory failure after extubation and had to be intubated again. Detailed medical history obtained from her relatives revealed she had an undiagnosed hoarseness for the last year. After this clinical clue we performed an electromyography. Nerve conduction study showed low amplitude Compound Muscle Activation Potential (CMAP) in left median and bilateral ulnar nerves, tibial CMAP amplitude much lower than distal; needle

EMG study showed severe acute denervation potentials, fasciculations and chronic degenerative Motor Unit Potentials (MUP). Acute denervation potentials and less often chronic neurogenic MUP's were seen in biceps muscle. Lower extremity muscles also showed acute denervation potentials. ALS diagnosis was made with these EMG findings in addition to clinical findings.Because our patient's symptoms were severe and in this stage Riluzol treatment is not very effective we applied ventilator support and administered antibiotherapy for the infections.The patient was hospitalized in our ICU connected to ventilator for 3 months and died because of pneumonia and sepsis.

Discussion

ALS is a progressive, relentless and destructive disease.Histopthological findings may be in different degrees in pyramidal beams, brain stem and spinal cord motor neurons. Generally ALS can be recognized easily by neurologists, however about 10% of the cases are misdiagnosed and thus delay is common [3].

The disease may present in different forms and during the course has several properties. Incidence is only about two cases per 100,000 populations in the United States that represents approximately 5.000 patients per year [4]. High prevalence of ALS seen in specific geographic areas in the Pacific Island of Guam (50 times more than western counties) sparked speculation about potential triggers such as environmental and genetic factors. There is no racial difference; men are affected almost twice as often as women. Although cases commonly have Mendelian autosomal dominant inheritance, it has also been found in autosomal recessive pattern [5].

Standard for ALS diagnosis at the present time is El Escorial diagnostic criteria [6].

Presence of:

1- Clinical, electrophysiological or histo-patological evidence of lower motor neuron involvement

2- Clinical evidence of upper motor neuron involvement

3- Medical history or physical examination suggesting progressive signs and symptoms within a specific body region or progression to the other regions

Absence of:

1- Electrophysiological or histo-patological data suggesting another disease that can explain lower or upper motor neuron involvement,

2- Imaging studies that can explain existing electrophysiological or histo-patological findings.

ALS onset is usually insidious and may present with unexplained motor disability of arm. Degree of pyramidal beam involvement in ventral horn is variable and not standard. Ocular muscles and urinary sphincter muscles are preserved. Cramps and fasciculation are frequent. Although ALS predominantly affects autonomic nervous system, spinocerebellar tract, the dorsal columns, extra motor cortex and the basal ganglia; there is growing evidence that it is a multisystemic disease [7].

Respiratory failure is most commonly a late stage symptom. Our patient diagnosed with ALS very late because she had muscle weakness in the lower extremities, but this was attributed to lumbar disc herniation and spinal stenosis and anybody didn't think of ALS.

ALS presentation with respiratory failure as first symptom is rare [8,9]. In such cases, patients often apply to pulmonary disease departments first and diagnosing ALS is extremely difficult. Bulbar onset, some patients have difficulty in swallowing and voice tonality changes. Respiratory function in the early stages of the disease is generally normal, shortness of breath occurs as the disease progresses. The symptoms occur during or after physical activity in early stages but as the disease progresses they also occur in the sitting position. Shortness of breath, shorter speech, the use of respiratory muscles and increase in the number of respiratory symptoms such as weak coughing are important evidences that show decrease in the respiratory muscles function [2].

In case of less obvious respiratory failure with a hint of a marked dyspnea, the patient suffers from insomnia. Patients can not sleep or wake up relaxed in the supine position often complain of headaches and excessive sleepiness during the day. If one or more of these symptoms present pulmonary function tests and blood gas sample should be implemented. If necessary, patients should be supported with noninvasive positive pressure ventilation devices. If these procedures are not enough and patients' respiration worsens tracheotomy

should be applied and breathing can be maintained artificially with the help of mechanical ventilation [4]. After ten days of intubation we applied tracheotomy to our patient.

Difficulty in communication is an important problem that ALS patients face. Communication may be through a panel that includes alphabetic letters. In our patient she had hoarseness for the last 1 year but was not diagnosed with anything.

Cause of ALS in most of the patients is still unknown. Treatment approaches targeting different pathogenic processes are emphasized [10]. Riluzol is a glutamate antagonist and has been used since 1996. It is the only agent that expands survival time. Daily dose of 100 mg Riluzol expands survival time of ALS patients approximately 3-5 months and is used in many countries. It has a medium efficacy, minor side effects and is expensive. There is an urgent need for new therapeutic agents that expands survival time. Current treatments target muscle proteins like 'Nogo', energy balance, cellular replacement and abnormal gene products resulted from mutations. There are two approaches on muscular level; change of proteins in neuromuscular junction like 'Nogo-A' or directly enhancing muscle strength [11]. Even though there are a couple hypotheses regarding its progression, etiology of the disease is not clarified exactly. There are studies that suggest stem cell therapies can be effective by targeting some of the mechanisms that are responsible for the etiology [12]. Because there is not any curative treatment for ALS, optimal treatment requires multidisciplinary approach aiming symptomatic control and maintenance of quality of life. Aerobic exercise and cognitive behavioral therapy are effective for maintaining and increasing functional capacity and enhance cognition respectively [13].

As a result; ALS is a rare disease, weakness in the voluntary muscles is in the foreground but ALS should also be kept in mind in the presence of respiratory distress and hoarseness.

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